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Evaluation of machine learning algorithms for improved risk assessment for Down's syndrome

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Abstract

Prenatal screening generates a great amount of data that is used for predicting risk of various disorders. Prenatal risk assessment is based on multiple clinical variables and overall performance is defined by how well the risk algorithm is optimized for the population in question. This article evaluates machine learning algorithms to improve performance of first trimester screening of Down syndrome. Machine learning algorithms pose an adaptive alternative to develop better risk assessment models using the existing clinical variables. Two real-world data sets were used to experiment with multiple classification algorithms. Implemented models were tested with a third, real-world, data set and performance was compared to a predicate method, a commercial risk assessment software. Best performing deep neural network model gave an area under the curve of 0.96 and detection rate of 78% with 1% false positive rate with the test data. Support vector machine model gave area under the curve of 0.95 and detection rate of 61% with 1% false positive rate with the same test data. When compared with the predicate method, the best support vector machine model was slightly inferior, but an optimized deep neural network model was able to give higher detection rates with same false positive rate or similar detection rate but with markedly lower false positive rate. This finding could further improve the first trimester screening for Down syndrome,

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